Newborn Screening Quality Assurance Program

PROFICIENCY TESTING | **Sickle Cell Disease and Other Hemoglobinopathies**

Volume 16, No. 1 | Quarter 1 | February 2006

INTRODUCTION

On January 9, 2006, we distributed to all active participants the Quarter 1 proficiency testing (PT) panel consisting of five dried-blood-spot (DBS) specimens for sickle cell disease and other hemoglobinopathies. A total of 75 PT panels were mailed by overnight FedEx mail. The packages went to 53 domestic laboratories and 22 foreign laboratories. The specimen panel consisted of five DBS specimens prepared from umbilical cord blood. This PT report is a compilation of all data reports for hemoglobinopathy testing received from participants by the designated deadline date. We distribute this quarterly report to all participants, state laboratory directors, and to program colleagues by request. We received data reports from 62 newborn screening laboratories. There were 13 laboratories that did not report this quarter. We requested that participants assay all survey specimens by the analytic schemes they routinely use and report for each specimen the presumptive phenotype, the presumptive clinical assessment, and any other clinical classifications that they deem consistent with their analytic results and program operations.

PARTICIPANTS' RESULTS

The certification report listing hemoglobins (Hbs) by phenotype and their presumptive clinical assessments appears on page 2.

The frequency distribution of reported phenotypes and presumptive clinical assessments appears on page 3.

The individual data verification for each laboratory with evaluation comments appears on page 4.❖

The NSQAP will ship next quarter's PT specimens on April 3, 2006.❖

SPOTLIGHT

Meetings

April 8-12, 2006, the 29th annual Meeting of the National Sickle Cell Disease Program will be held in Memphis, TN, at the Peabody Hotel. The program will be hosted by the Comprehensive Sickle Cell Center at St. Jude Children's Research Hospital. For more information visit us at http://www.stjude.org/sicklecell-conf.❖

News

The January 2006 issue of Nature Biotechnology published a study where scientists from Memorial Sloan-Kettering Cancer Center have devised a novel strategy that uses stem cell-based gene therapy and RNA interference to genetically reverse sickle cell disease (SCD) in human cells. The study describes how a viral vector was introduced into adult stem cell cultures of SCD patients. The vector carries a therapeutic globin gene harboring an embedded small interfering RNA precursor designed to suppress abnormal hemoglobin formation. Researchers found that the newly formed red blood cells made normal hemoglobin and suppressed production of the sickle shaped hemoglobin typical of the disease.❖

ACKNOWLEDGMENTS

The specimens for this survey were prepared from umbilical cord blood samples supplied by Alabama State Public Health Laboratory.❖

CDC/APHL

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CENTERS FOR DISEASE CONTROL AND PREVENTION

Newborn Screening Quality Assurance Program
Sickle Cell Disease and Other Hemoglobinopathies

Specimen and Lab Certification

Year: 2006 Quarter: 1

Presumptive Clinical Phenotypes

	Specimen 1631	Specimen 1632	Specimen 1633	Specimen 1634	Specimen 1635
Expected Presumptive Phenotype	FAS	FAC	FA	FS	FAS
Accepted Presumptive Phenotypes	FSA	FCA			FSA

Presumptive Clinical Assessments

	Specimen 1631	Specimen 1632	Specimen 1633	Specimen 1634	Specimen 1635
Expected Presumptive Clinical Assessment	02	03	01	04	02
Accepted Presumptive Clinical Assessments					

01 Normal--no abnormal Hb found
02 Hemoglobin S carrier
03 Hemoglobin C carrier
04 Hemoglobin SS disease (Sickle cell anemia)
05 Hemoglobin SC disease
06 Hemoglobin SD disease
08 Hemoglobin D carrier
09 Hemoglobin E carrier
12 Hemoglobin S, E disease
16 Alpha-thalassemia (Bart's Hb)
18 Hemoglobin EE disease
20 Assessment not listed
21 Unsatisfactory specimen
22 Unidentified variant, fast or aging band
Specimen not evaluated (NE)

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Frequency Distributions

Year: 2006 Quarter: 1

Phenotypes

Specimen Number	Hemoglobin Phenotypes	Frequency Distributions
1631	FAS	62
1632	FA	1
	FAC	61
1633	FA	62
1634	FS	61
	FSS	1
1635	FAC	1
	FAS	61

Clinical Assessments

Specimen Number	Presumptive Assessments	Frequency Distributions
1631	02 Hemoglobin S carrier	62
1632	01 Normal	1
	03 Hemoglobin C carrier	61
1633	01 Normal	62
1634	04 Hemoglobin S, S disease (Sickle cell anemia)	62
1635	02 Hemoglobin S carrier	62

This *NEWBORN SCREENING QUALITY ASSURANCE PROGRAM* report is an internal publication distributed to program participants and selected program colleagues. The laboratory quality assurance program is a project cosponsored by the **Centers for Disease Control and Prevention (CDC) and the Association of Public Health Laboratories.**

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